



*Case Report*

## **A novel mutation of the MFN2 gene in a Chinese family with Charcot-Marie-Tooth disease**

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**ABSTRACT.** Charcot-Marie-Tooth (CMT) is a group of clinically and genetically heterogeneous inherited neuromuscular disorders. At present, more than 30 loci have been reported to be associated with CMT disease; point mutations in the mitofusin 2 (MFN2) gene is one of the most common causes. We studied a Chinese family with CMT disease in which the phenotype of affected individuals varied, and the weakness condition of the distal legs in males, except the proband, was less severe than in females in this family. Linkage analysis and PCR sequencing revealed a missense mutation (NM\_014874.3:c.1066 A>G) in the MFN2 gene, resulting in an amino acid substitution of threonine to alanine in condon 356 (Thr356Ala). This is a novel phenotype and mutation for CMT family.

**Key words:** Charcot-Marie-Tooth; Mitofusin 2; Mutation; Gender